1 Haemoglobin is a protein found in red blood cells that helps transport oxygen in the blood.

In sickle cell anaemia, the presence of the allele (**a**) in place of the normal allele (**A**) results in a change of one amino acid in the haemoglobin molecule.

This mutation changes the properties of haemoglobin and can result in red blood cells becoming less flexible and blocking small blood vessels.

The frequency of the recessive allele (**a**) is much higher in populations in West Africa than in populations in Northern Europe.

People in West Africa have a much higher risk of being infected with malaria parasites and developing severe anaemia than people in Northern Europe.

People who are heterozygous for sickle cell anaemia have sufficient amounts of normal haemoglobin to prevent severe anaemia.

One theory suggests that the malaria parasite (*Plasmodium sp.*) causes red blood cells with any modified haemoglobin to rupture. This occurs before the parasite can reproduce.

*(a) In West Africa, the average life expectancies of people with genotypes **AA**, **Aa** or **aa** are different.

(i) Using the information above, give an explanation for the difference in life expectancy of people who do not have the recessive allele.

(2)

(ii) Using the information above, give an explanation for the difference in life expectancy of people in West Africa who have the genotype **Aa**.

(2)

*(b) Explain how a change of one amino acid can lead to a change in the structure and properties of the haemoglobin protein.

(4)

(Total for Question 1 = 8 marks)

2 The scientific article you have studied is adapted from the book called The Immortal Life of Henrietta Lacks by Rebecca Skloot. Published by Pan Books in 2011.

(a) Explain what is meant by the term **mitosis** (paragraph 7).

(2)

(b) The genome makes sure that cells 'do their jobs, whether that's controlling your heartbeat or helping your brain understand the words on this page' (paragraph 10).

Describe how cells in the sino-atrial node (SAN) are involved in controlling heart rate.

(3)

(c) Henrietta's cells have 'been used to study lactose digestion' (paragraph 16).

Suggest how her cells may have digested lactose.

(3)

(d) 'Like guinea pigs and mice, Henrietta's cells have become the standard laboratory workhorse' (paragraph 16).

Suggest **two** reasons why it is preferable to use Henrietta's cells in medical research, rather than using guinea pigs and mice.

(2)

*(e) 'By the end of 1951, the world was in the midst of the biggest polio epidemic in history' (paragraph 19). This was caused by poliovirus which can lead to paralysis (paragraph 20).

The virus infects motor neurones which can stop skeletal muscles from working.

Suggest how an infection of motor neurones by the virus can stop the transmission of nerve impulses and lead to muscle paralysis.

(6)

(f) Poliovirus, like Human Immunodeficiency Virus, is a retrovirus. Poliovirus was able to infect HeLa cells (paragraph 25).

Give **three** similarities between the structure of the genetic material in poliovirus and the genetic material in HeLa cells.

(3)

(g) Scientists had studied genes by breeding plants 'then breeding their offspring to see how genetic traits are passed from one generation to the next' (paragraph 33).

When this was done using a smooth pea and a wrinkled pea, it was found that in the F2 generation (second generation of offspring), 75% were smooth.

In the space below, draw genetic diagrams to describe and explain the genotypes of the parents and their offspring in the previous **two** generations.

(4)

(h) Explain what is meant by the term **human genome map** (paragraph 37).

(2)

(i) Suggest how the 'p53 tumor suppressor gene' (paragraph 43) could stop a potential tumour cell forming.

(2)

(j) Using paragraph 46, suggest what the 'specific DNA sequence from a blood cell' coded for.

(2)

(k) A human telomere (paragraph 60) contains 10 000 nucleotides. Using information from paragraph 58, state the number of telomere nucleotides lost per cell division.

(1)

Answer ..... nucleotides

(Total for Question 2 = 30 marks)